

EA2 – kirjallisuusviitteet

- Griggs RC et al., Hereditary paroxysmal ataxia: response to acetazolamide. *Neurology* 28: 1259 – 1264, 1978
- Bressman S: Genetics of movement disorders: recent advances. Kokousyhteenvedossa: Movement disorders, s. 440-67 – 440-102. American Acadey of Neurology, 48th Annual Meeting, March 23 – 30, San Fransisco, 1996
- Damji KF et al., Periodic vestibulocerebellar ataxia, an autosomal dominant ataxia with defective smooth pursuit, is genetically distinct from other autosomal dominant ataxias. *Arch Neurol* 53: 338 – 344, 1996
- Baloh RW et al., Familial episodic ataxia: clinical heterogeneity in four families linked to chromosome 19p. *Ann Neurol* 41: 8 – 16, 1997
- Brandt T and Strupp M: Episodic ataxia type 1 and 2 (familial periodic ataxia/vertigo). *Audiol Neurotol* 2 (6): 373 – 383, 1997
- Furman JM: Otolith-ocular responses in familial episodic ataxia linked to chromosome 19p. *Ann Neurol* 42: 189 – 193, 1997
- Meisler MH et al., Ion channel mutations in mouse models of inherited neurological disease. *Ann Med* 29: 569 – 574, 1997
- Kaakkola S ja Rinne R: Ataksiat ja niiden erotusdiagnostiikka. *Duodecim* 113: 1773 – 1782, 1997
- Somer H ja Auranen M: Lihastautien geenivirheet. – katsaus. *Duodecim* 113: 1795 – 1802, 1997
- Barchi RL: Ion channel mutations affecting muscle and brain. *Curr Opin Neurol* 11: 461 – 468, 1998
- Gordon N: Episodic ataxia and channelopathies. *Brain&Development* 20: 9 – 13, 1998
- Subramony SH and Nance M: Diagnosis and Management of the inherited ataxias. *The Neurologist* 4: 327 – 338, 1998
- Terwindt GM et al., Migraine, ataxia and epilepsy: a challenging spectrum of genetically determined calcium channelopathies. Dutch Migraine Genetics Research Group. *Eur J Hum Genet* 6 (4): 297 – 307, 1998
- Davies NP and Hanna MG: Neurological channelopathies: diagnosis and therapy in the new millennium. *Ann Med* 31: 406 – 420, 1999
- Denier C et al., High prevalence of CACNA1A truncations and broader clinical spectrum in episodic ataxia type 2. *Neurology* 52 (9): 1816 – 1821, 1999
- Ducros A et I., Recurrence of the T666M calcium channel CACNA1A gene mutation in familial hemiplegic migraine with progressive cerebellar ataxia. *Am J Hum Genet* 64 (1): 89 – 98, 1999
- Randall A and Benham CD: Recent advances in the molecular understanding of voltage-gated Ca²⁺channels. *Neuroscience* 14: 255 – 272, 1999
- Sappey-Marinier D et al., Phosphorous and proton magnetic resonance spectroscopy in Episodic Ataxia type 2. *Ann Neurol* 46: 256 – 259, 1999

Escayg A et al., Coding and noncoding variation of the human calcium-channel beta4-subunit gene CACNB4 in patients with idiopathic generalized epilepsy and episodic ataxia. Am J Hum Genet 66: 1531 – 1539, 2000

Baloh RW: Periodic and progressive ataxias, sivut 229 - 238. Kirjassa: Channelopathies of the Nervous System. Toim. Rose MR ja Griggs RC. Butterworth and Heinemann, 2001

Denier C et al., Missense CACNA1A mutation causing Episodic Ataxia type 2. Arch Neurol 58: 292 – 295, 2001

Goadsby PJ and Ferrari MD: Migraine: a multifactorial, neurovascular episodic channelopathy?, sivut 274 – 292. Kirjassa: Channelopathies of the Nervous System. Toim. Rose MR ja Griggs RC. Butterworth and Heinemann, 2001

Guida S et al., Complete loss of P/Q calcium channel activity caused by a CACNA1A missense mutation carried by patients with episodic ataxia type 2. Am J Hum Genet 68 (3): 759 – 764, 2001

Jen J (Ed): Ataxia and Calcium Channels. What a Headache! Arch Neurol 58: 179 – 180, 2001

Jen J et al., Loss-of-function EA2 mutations are associated with impaired neuromuscular transmission. Neurology 57(10): 1843 – 1848, 2001